

**Long Term Plan
for
Genetics
in the
Department of Health**

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Introduction

As the number of deaths due to infectious or environmental causes decrease, the proportion of deaths due to genetic disease increase. Also, the Human Genome Project, which maps all the human genes, will be completed in the next decade. This makes genetics the important focus for current and future public health issues. In accordance with the Institute of Medicine's, The Future of Public Health (1988), the long term plan for genetics in the Department of Health (Department) must include components for assessment, policy development and assurance.

Assessment

Assessment involves identifying and defining the genetic needs of the community. Continued assessment also provides information about the effectiveness of policies and services which are implemented. The first genetics needs assessment for Hawaii was done in 1978 by a public health graduate student at the University of Hawaii. This assessment found Hawaii lacking in all areas of genetics. It was 15 years before another genetics needs assessment was done. In 1993, the Pacific Southwest Regional Genetics Network allotted a portion of its federal grant to perform this needs assessment. The final document was made available in November 1993. This time the assessment showed that Hawaii had improved its genetic services but there are still some areas that either need more improvement or are non-existent. Many of the recommendations from the current needs assessment will be included in this long term plan.

Policy Development

In the past, policy development for genetics in the Department has been very fragmented. Policies have been set in a variety of ways, but the major problem is that the intent of the policies is sometimes lost with personnel changes. Another complaint is that some policies are made without private clinical or community consultation. Currently, we try to obtain community involvement through the many advisory committees when policies are evaluated or developed. The development of the State Genetics Advisory Committee consisting of public and private sector members will encourage cooperative policy development. The committee will play a major role in advising the Department on policies, programs and activities which reflect the standards and principles for genetics we want in the Hawaii.

Assurance

The Department needs to play a leadership role in activities that promote the prevention, treatment and rehabilitation of genetic disorders. This involves coordinating services, education, and technical assistance. The Department also has an important role in promoting and in some circumstances, providing mechanisms for quality control for all aspects of genetic services and education. In these times of reduced budget, it is even more important for the Department to work with the community to assure that quality genetic activities and services will be available.

Plan

1) State Genetics Advisory Committee

In order to coordinate the genetic activities in Hawaii and make genetics more community based, a State Genetics Advisory Committee was formed in May 1995. This committee will have representatives from the Department of Health, clinicians, private laboratories, educators and the lay community. This committee will be responsible for advising the Department on its genetic policies, programs and activities. The main committee will also be expected to participate in planning activities for the Pacific Southwest Regional Genetics Network (PSRGN) sub-contract.

Newborn Screening Program and Pacific Southwest Regional Genetics Network-Hawaii sub-committees have been formed. We are also investigating the feasibility of bringing the Birth Defects Monitoring Program Advisory sub-committee under the main committee. Ad hoc sub-committees can also be formed when specific genetic issues need to be addressed and then the report from the sub-committees can be presented to the main committee for adoption.

2) Newborn Screening Program

The genetics section should continue to investigate the feasibility of collecting fees for the Newborn Screening (NBS) Program services. The fees can pay for a contract with one central laboratory for all the newborn screening tests, follow-up of positive screens, and education. Centralizing laboratory services will simplify the NBS program's laboratory quality assurance role. The fees will also allow the Department to increase the number of disorders screened. This method of fee collection is successful in many states including Nevada which has a birth rate comparable to Hawaii. Ultimately, a centralized newborn screening program will result in less expensive, but higher quality, testing for the public.

We are investigating the addition of other newborn screening tests to our current phenylketonuria and congenital hypothyroid testing. Hawaii is one of two states (South Dakota is the other one) that only screens for two disorders. All other states screen for 3 or more disorders.

The NBS Program also needs better data collection systems. We need to update the current computer programs and linkages in order to transfer information electronically rather than manually. This would save time which is important for the confirmation of a positive screening result and initiation of treatment as rapidly as possible.

Of course the NBS program should continue to provide education, encourage public/private linkages, provide quality assurance of tests and follow-up services, and participate in policy and legislation development for newborn screening.

3) Data collection

The private and public sectors have several databases which contain genetic information. This arrangement does not allow sharing of information or proper data analysis. In many cases the same information is duplicated in several databases. A coordinated and cooperative database system may allow tracking of utilization, effectiveness of services and help identify gaps in service provision. We are currently investigating new technologies to aid data collection (eg. optical character scanning).

The Department must address the problem of coordinating the databases without jeopardizing confidentiality of the information. As more genetic tests become available, records of these tests will be kept in many databases. The Department needs to play a leadership role in developing policies to address the legal aspects of access to genetic information. The State Genetics Advisory committee can play a major role in arranging a cooperative effort in database coordination and a discussion about the legal and ethical aspects regarding the information in the databases.

4) Birth Defects Monitoring Program

The Department should promote the importance of the Birth Defects Monitoring Program. The accurate data obtained from this program allows good statistical analysis and assessment of genetic disorders in Hawaii. Without this data, analysis of the effectiveness of genetic programs may not be accurately calculated.

The Department will work with the program to determine a more cost-effective means to collect birth defects data. We are also investigating the overlaps in the data collection system in order to possibly streamline the data collection.

5) Prenatal Screening

The Prenatal Screening (PNS) Program has been monitoring Maternal Serum Alpha-Fetoprotein (MSAFP) testing for the past six years. The PNS program does not provide any direct intervention or follow-up services for the women who have MSAFP screening. Currently, a new test called, Multiple Marker Screening (Triple Screening), is replacing MSAFP testing. The Multiple Marker Screening can detect 40% more cases of fetal Down syndrome than MSAFP testing.

The PNS program should concentrate its resources on education. The statistics for FY1994 in Hawaii show that approximately 75% of eligible pregnant women have MSAFP or Multiple Marker screening. The statistics also show that approximately 20-25% of pregnant women in Hawaii do not present for early prenatal care. This is the same population who do not receive prenatal screening since the screening test must be done before 21 weeks of pregnancy. A cooperative educational effort with the Perinatal

Section of the Maternal and Child Health Branch will help increase the number of women offered prenatal screening in Hawaii.

The PNS program should also encourage linkages with the Birth Defects Monitoring Program (BDMP) and laboratories. We are currently participating in a pilot project with the BDMP to assess the results when the two program's data are cross-matched. The program also has had great success in bringing the private laboratories together to discuss prenatal screening. This activity should continue since it benefits all the laboratories and the community.

6) General Genetic Screening

The rapid advances in genetics has allowed the development of greater numbers of genetic screening tests. A good screening test will determine those at greatest risk of having a genetic predisposition or disorder so they can be referred to more accurate, but expensive, diagnostic testing. After a presumptive positive test, a diagnostic test is required to accurately determine your risk for the disorder. The Department must play a leadership role in the evaluation, education and promotion of quality genetic screening tests. This will be a continuing role since genetic screening tests change rapidly.

The Department will also have to take a major role in dealing with the legal and ethical aspects of genetic screening. The Human Genome Project is scheduled for completion in the next ten years. The amount of potential information one can gain from screening tests will only increase. The Department, in a cooperative effort with other State agencies and the community, must develop policies to deal with the ethical and legal aspects of this explosion of information.

7) Clinical and Support Services

An important aspect of genetic disorders is that there are currently no cures. Treatment is available for many genetic disorders but the basic genetic defect is unchanged. The Department needs to recognize that genetic disorders are chronic and can initially manifest at anytime in a person's life. Therefore, clinical and support services are very important for the prevention, treatment and rehabilitation of genetic disorders. The Department has the responsibility for coordinating and encouraging the private sector to provide quality genetic services. At the very least, the Department should act as a referral source for available genetic services in Hawaii.

8) Education

Continuing education is a very important component of the long term plan for genetics in this State. Education will provide the groundwork as the Human Genome Project nears completion. It will also promote prevention and high quality genetic health care by increasing genetic awareness.

A genetics speakers bureau has been formed as part of the educational effort. Volunteers from the genetics community have agreed to speak on a variety of genetic subjects. The Genetics Advisory Committee or the Department can screen the lecturers and/or subjects. The availability of the speakers bureau will be publicized to the community. The Department can coordinate the lecturers/lectures and also fund neighbor island travel, if funds are available.

The Department should also continue to co-sponsor genetics educational efforts with the private sector. This has worked well in the past with the PSRGN and March of Dimes. This will also help promote coordinated genetic service delivery.

The Department will try and continue to publish newsletters as funds permit. However, instead of many newsletters for the different programs, one State Genetics Newsletter should be created. This will stop the delivery of fragmented information and promote genetic information under one umbrella. One newsletter will also reduce mailing costs.

9) Funding

The present programs which involve genetics are being assessed to determine if restructuring can produce increased effectiveness without increased funding. A cooperative effort between programs may allow better educational efforts, data collection and dissemination of information. There will also be a better determination of community needs when all the programs work together. This collaborative effort will be helpful when requesting grant funding.

The Department should continue to encourage private, federal and state funding for genetic services as needs are identified. Also, funding should be obtained to continue the development of the infrastructure for genetics. The funding can come from federal granting agencies, the state legislature or private agencies. These resources are currently being investigated.

The Department should also play a strong leadership role in assessing and promoting insurance coverage for the prevention, testing, treatment and rehabilitation for genetic disorders. Activities at the national level to develop CPT codes and reasonable reimbursement rates for genetic services will aid our efforts.

10) Legal and Ethical Issues

The Department of Health, in cooperation with other State agencies, will have to address the legal and ethical implications of genetic testing. The lack of controls over a person's genetic information can lead to discrimination (personally and professionally) and loss of health care coverage.

Seven states have enacted legislation to prevent discrimination by health insurance companies by prohibiting denial of coverage due to genetic disease or the risk of genetic disease. We are currently assessing the situation in Hawaii to see if we need to draft similar legislation. We are also monitoring the legislative bills being introduced with respect to the legal and ethical issues relating to genetic disorders.

11) Quality Assurance

In order to play a leadership role in promoting and developing policies to maintain the level of genetic services in Hawaii, the Department must keep current on the changes in genetic information and technology. This will be a difficult task since the doubling time for genetic information is approximately two years. Therefore, we must work cooperatively with the experts in the community to advise us as new information arises.

The Department should act as an objective body to promote and/or monitor quality assurance. To evaluate the quality of genetic services in the State, the Department must first determine the minimal national standards. Then the Department must use its assessments to compare the actual quality of genetic services in Hawaii with national standards. Deficiencies should be noted and changes recommended or in some cases, legislated. The Department may also determine if efforts at other levels (eg. federal regulatory agencies, funding agencies, insurance, professional organizations) should be involved in determining changes. Follow-up will need to be done to determine the effectiveness of the changes and if more changes need to occur.

Another major role for the Department will be coordinating education and training efforts to familiarize the clinicians, laboratories and the public with the changes. The Department may also have to allocate resources towards this effort.

Role of the State Genetics Coordinator

The State Genetics Coordinator will be expected to play the primary role in initiating and coordinating each of the activities in the long term plan. It is important for the Coordinator to be consistently involved in planning, developing and implementing programs and policies involving genetics. This continuity will help ensure that the public and private sectors are working in a cooperative and effective effort. This relationship will also allow the Department to play a strong leadership role in genetics since the Coordinator will be expected to anticipate changes and trends in genetics and plan a program of action for the Department.